

20220203_data_release_notes

Overview of ClinVar submitted records: 2022

Jan 01, 2022	1,895,787
Feb 01, 2022	1,926,598

Overview of changes in the ClinVar release of February 3, 2022

Content

Brief	Explanation
ClinGen Malignant Hyperthermia Susceptibility Variant Curation Expert Panel (Organization ID 508072)	ClinGen Malignant Hyperthermia Susceptibility Variant Curation Expert Panel submitted 61 novel variant interpretations and updates to 8 records.
ClinGen Platelet Disorders Variant Curation Expert Panel (Organization ID 507662)	ClinGen Platelet Disorders Variant Curation Expert Panel submitted 49 novel variant interpretations and updates to 2 records.
ClinGen Monogenic Diabetes Variant Curation Expert Panel (Organization ID 508240)	ClinGen Monogenic Diabetes Variant Curation Expert Panel submitted 31 novel variant interpretations and updates to 3 records.
ClinGen Rett and Angelman-like Disorders Variant Curation Expert Panel (Organization ID 508065)	ClinGen Rett and Angelman-like Disorders Variant Curation Expert Panel submitted 1 novel variant interpretation.
ClinGen TP53 Variant Curation Expert Panel (Organization ID 507142)	ClinGen TP53 Variant Curation Expert Panel submitted updates to 4 records.
CeGaT Praxis fuer Humangenetik Tuebingen (Organization ID 505870)	CeGaT Praxis fuer Humangenetik Tuebingen submitted updates to 28,115 records.
Genetic Services Laboratory, University of Chicago (Organization ID 1238)	Genetic Services Laboratory, University of Chicago submitted 8,254 novel variant interpretations and updates to 204 records.
Nilou-Genome Lab (Organization ID 507598)	Nilou-Genome Lab submitted 3,974 novel variant interpretations.
ARUP Laboratories, Molecular Genetics and Genomics (Organization ID 25969)	ARUP Laboratories, Molecular Genetics and Genomics submitted 2,659 novel variant interpretations and updates to 3,387 records.
Color Health, Inc (Organization ID 505849)	Color Health, Inc submitted 1,905 novel variant interpretations and updates to 12,835 records.
3billion (Organization ID 507830)	3billion submitted 1,080 novel variant interpretations and updates to 6 records.
GeneDx (Organization ID 26957)	GeneDx submitted 921 novel variant interpretations and updates to 834 records.
Greenwood Genetic Center Diagnostic Laboratories (Organization ID 1019)	Greenwood Genetic Center Diagnostic Laboratories submitted 787 novel variant interpretations and updates to 15 records.

Women's Health and Genetics/Laboratory Corporation of America, LabCorp (Organization ID 500026)	Women's Health and Genetics/Laboratory Corporation of America, LabCorp submitted 758 novel variant interpretations and updates to 94 records.
Centogene AG - the Rare Disease Company (Organization ID 279559)	Centogene AG - the Rare Disease Company submitted 664 novel variant interpretations.
Broad Institute Rare Disease Group, Broad Institute (Organization ID 506627)	Broad Institute Rare Disease Group, Broad Institute submitted updates to 663 records.
Genome Diagnostics Laboratory, The Hospital for Sick Children (Organization ID 1043)	Genome Diagnostics Laboratory, The Hospital for Sick Children submitted 540 novel variant interpretations.
Neuberg Supratech Reference Laboratories Pvt Ltd, Neuberg Centre for Genomic Medicine (Organization ID 508108)	Neuberg Supratech Reference Laboratories Pvt Ltd, Neuberg Centre for Genomic Medicine submitted 465 novel variant interpretations.
Myriad Women's Health, Inc. (Organization ID 507240)	Myriad Women's Health, Inc. submitted 391 novel variant interpretations and updates to 12 records.
Kasturba Medical College, Manipal, Manipal Academy of Higher Education (Organization ID 505991)	Kasturba Medical College, Manipal, Manipal Academy of Higher Education submitted 341 novel variant interpretations and an update to 1 record.
GeneOne, DASA (Organization ID 508087)	GeneOne, DASA submitted 158 novel variant interpretations.
Coming soon: new values of clinical significance for variants with low penetrance and risk alleles	<p>We are adding new terms for the clinical significance (interpretation) of variants with low penetrance and risk alleles, based on this white paper from ClinGen:</p> <p>https://clinicalgenome.org/site/assets/files/4531/clingenrisk_terminology_recomendations-final-02_18_20.pdf</p> <p>The new terms are:</p> <ul style="list-style-type: none"> • Pathogenic, low penetrance • Likely pathogenic, low penetrance • Established risk allele • Likely risk allele • Uncertain risk allele <p>We are also updating how we report a list of values in the aggregate clinical significance when there are terms on the scale from pathogenic to benign and other kinds of terms like drug response. These terms will be listed with a semi-colon delimiter, instead of a comma, to accommodate some of the new terms that include a comma. The aggregate clinical significance is represented in the VCV XML by the element InterpretedRecord/Interpretation/Description, and in the VCF file as the CLNSIG info tag.</p> <p>e.g. for Variation ID 48692, the aggregate clinical significance will change from</p> <p>Pathogenic, drug response</p> <p>to</p> <p>Pathogenic; drug response</p> <p>These changes are planned for a late February deployment, so they are expected to be in the next monthly release.</p>

Overview of submitted records: 2021

Jan 01, 2021	1,325,194
Feb 01, 2021	1,351,093
Mar 01, 2021	1,353,834
Apr 01, 2021	1,419,210

May 01, 2021	1,441,450
Jun 01, 2021	1,517,898
July 01, 2021	1,602,096
Aug 01, 2021	1,624,873
Sep 01, 2021	1,672,649
Oct 01, 2021	1,809,253
Nov 01, 2021	1,847,096
Dec 01, 2021	1,872,380

Overview of submitted records: 2020

Jan 01, 2020	1,026,969
Feb 01, 2020	1,041,077
Mar 01, 2020	1,055,499
Apr 02, 2020	1,084,731
May 01, 2020	1,136,163
Jun 01, 2020	1,142,645
Jul 01, 2020	1,255,451
Aug 01, 2020	1,302,013
Sep 01, 2020	1,307,377
Oct 01, 2020	1,310,477
Nov 01, 2020	1,315,943
Dec 03, 2020	1,322,303

Overview of submitted records: 2019

Jan 01, 2019	759562
Feb 07, 2019	778673
Mar 01, 2019	782638
Apr 01, 2019	787656
May 01, 2019	795045
Jun 01, 2019	811551
Jul 01, 2019	819827
Aug 01, 2019	825177
Sept 01, 2019	881419
Oct 01, 2019	888298
Nov 01, 2019	889968
Dec 01, 2019	893196

Overview of submitted records: 2018

Jan 01, 2018	579543
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Feb 01, 2018	582113
Mar 01, 2018	593651
Apr 01, 2018	610005
May 01, 2018	645149
Jun 01, 2018	676018
Jul 01, 2018	676575
Aug 01, 2018	685942
Sep 01, 2018	701880
Oct 01, 2018	708726
Nov 01, 2018	715516
Dec 01, 2018	749203

Overview of submitted records: 2017

Jan 01, 2017	396005
Feb 01, 2017	405182
Mar 01, 2017	406220
Apr 01, 2017	446265
May 01, 2017	482941
Jun 01, 2017	486420
Jul 01, 2017	488658
Aug 01, 2017	492592
Sep 01, 2017	504299
Oct 01, 2017	512373
Nov 01, 2017	517157
Dec 01, 2017	519359

Overview of submitted records: 2016

Jan 01, 2016	172867
Feb 01, 2016	176710
Mar 01, 2016	178032
Apr 01, 2016	180549
May 01, 2016	181155
Jun 01, 2016	192617
Jul 01, 2016	204415
Aug 01, 2016	209842
Sep 01, 2016	210200
Oct 01, 2016	213499
Nov 01, 2016	236420

Dec 01, 2016	240042
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Overview of submitted records: 2015

Jan 01, 2015	149013
Feb 01, 2015	156999
Mar 01, 2015	162455
Apr 01, 2015	171408
May 01, 2015	172044
Jun 01, 2015	173236
Jul 01, 2015	184506
Aug 01, 2015	154686
Sep 01, 2015	158580
Oct 01, 2015	160538
Nov 01, 2015	170931
Dec 01, 2015	172006

Overview of submitted records: 2014

Jan 01, 2014	68204
Feb 01, 2014	73492
Mar 01, 2014	83343
Apr 01, 2014	111501
May 01, 2014	112349
Jun 01, 2014	117209
Jul 01, 2014	127132
Aug 01, 2014	127557
Sep 1, 2014	143114
Oct 1, 2014	143601
Nov 1, 2014	144117
Dec 1, 2014	148008

Overview of submitted records: 2013

Apr 05, 2013	30333
May 01, 2013	30386
Jun 01, 2013	39047
Jul 01, 2013	39170
Aug 01, 2013	45901
Sep 01, 2013	50263
Oct 01, 2013	52047

Nov 01, 2013	64750
Dec 01, 2013	64881